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Case Report Section

t(4;11)(q23;p15) in paediatric early T cell precursor acute lymphoblastic leukemia

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Abstract

Case report on t(4;11)(q23;p15) in paediatric early T cell precursor acute lymphoblastic leukemia.

Clinics

Age and sex
12 years old female patient.

Previous history
No preleukemia, no previous malignancy, no inborn condition of note

Organomegaly
Hepatomegaly (3 cms below right costal margin), splenomegaly (3 cms below left costal margin), enlarged lymph nodes (Cervical lymphadenopathy), no central nervous system involvement

Blood

WBC: 1.49X 10^9/l
HB: 6.6g/dl
Platelets: 1.9X 10^9/l
Blasts: 2%
Bone marrow: 30%; Hypocellular and aparticulate bone marrow aspirate smears and show presence of 30% blasts.

Cyto-Pathology Classification

Phenotype: L1

Immunophenotype
The blast cells were dim CD45 positive and expressed moderate CD34, CD38, CD7, CD33, dim CD2, partially CD117 and dim cytoplasmic CD3. These cells are negative for CD19, CD10, CD20, HLADR, CD4, CD8, surface CD3, CD5 and CD1a.

Rearranged Ig Tcr
Not done

Pathology
Acute lymphoblastic leukemia

Electron microscopy
Not done

Diagnosis
Early T cell precursor Lymphoblastic leukemia.

Survival

Date of diagnosis
12-2015

Treatment
High risk BFM 95

Complete remission
Relapse: no

Status Alive

Last follow up
11-2017

Survival
24+months

Karyotype

Sample Bone marrow aspirate

Culture time 17 overnight with and without colcemid

Banding GTG

Results
46,XX,t(4?11)(q23?p15)[09]/46,XX[11]

Other molecular cytogenetics technics
Fluorescence in situ hybridisation was done on bone marrow culture pellet using NUP98 break apart probe, Zytovysion, Fischai, Bremerhaven, Germany.

Other molecular cytogenetics results
Positive for rearrangement of the NUP98 gene in 34% of the cells.

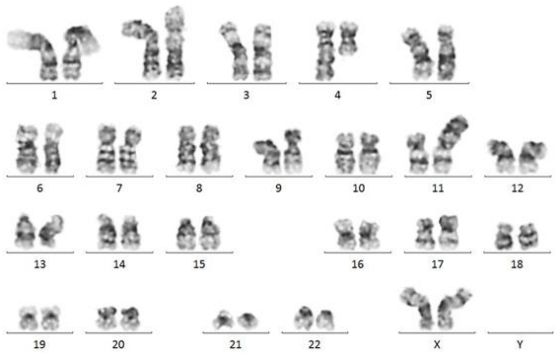

Figure 1: Karyogram showing t(4;11)(q23?p15) GTG staining and banding method × 100 oil immersion, Carl Zeiss Axios cope and processed using IKAROS software)

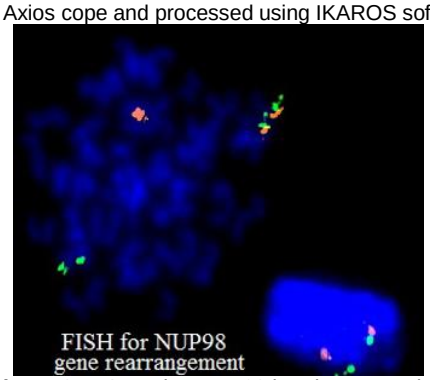

Figure 2: FISH using NUP98 break apart probe, Zytovysion, Fischai, Bremerhaven, Germany.

Other Findings

Negative for BCR/ABL1 fusion, ETV6/RUNX1 fusion, KMT2A gene rearrangement and E2A gene rearrangement by FISH.

Comments

The t(4;11)(q23;p15) is a rare recurrent translocation in T cell acute lymphoblastic leukemia (T cell ALL) that has been described in ten cases till date and leads to the fusion of NUP98 gene and RAP1GDS1 gene (Mohamed AN, In press). The uniqueness of this translocation is it not involving the T cell receptor genes. It has previously been associated with T cell lymphoblastic leukemias arising from progenitor cells aberrantly expressing myeloid molecules (Mecucci, C. et.al., 2000). In the present case this translocation was detected in early precursor T cell lymphoblastic leukemia (EPT-ALL), a recently described sub entity of T cell ALLs associated with inferior outcome that is characterised by CD1a negative, CD8 negative, CD5 weak, aberrant expression of myeloid and or stem cell antigens like CD117 CD13 and CD33 (Coustain S. et.al., 2009). Although the previous published three case reports described aberrant myeloid antigen expression, none were reported with CD117 expression. Nine cases were of adults while a single case was described in 6 years old female. The translocation was detected at diagnosis in eight cases and was associated with additional abnormalities in three cases. In one case it was cytogenetically cryptic and was detected by molecular studies (Cimino G. et.al., 2001). The t(4;11) is associated with unfavourable outcome (Mohamed AN, In press). All the described cases died of the disease except a 6 years old female child who remained alive after 25 months of follow up (Pui CH. et.al., 1991). The present case was of EPTALL with t(4;11) who is alive and remains in remission 24 months post diagnosis. It is postulated that t(4;11) may impart unfavourable prognosis in adults rather than paediatric population; however, further studies are warranted.

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